



**Fanconi Anemia /  
Constitutional Anemia  
Pre-HSCT Data**

**Registry Use Only**

Sequence  
Number:

Date  
Received:

CIBMTR Center Number:

CIBMTR Recipient ID:

Today's Date:          
Month Day Year

Date of HSCT for which this form is  
being completed:          
Month Day Year

HSCT type:  autologous  allogeneic,  allogeneic,  syngeneic  
unrelated related (identical twin)

Product type:  marrow  PBSC  cord blood  other product,  
specify: \_\_\_\_\_

**This form must be accompanied by Form 2000 – Recipient Baseline Data. All information in the box above, including the date, should be identical with the corresponding Form 2000. Information should come from an actual examination by the Transplant Center physician, or the physician who is following the recipient pre-HSCT, or abstraction of the recipient’s medical records.**

**If this is a report of a second or subsequent transplant, check here  and continue with question 153.**

1. What was the date of diagnosis of Fanconi Anemia?          
Month Day Year

2. Was the diagnosis made in utero?  
 1  yes  
 2  no

Specify test(s) performed to identify disease:

3. 1  yes 2  no Amniocentesis

4. 1  yes 2  no Chorionic villous sampling (CVS)

5. 1  yes 2  no Fibroblasts

6. 1  yes 2  no Other test

7. If yes, specify other test: \_\_\_\_\_

8. Was the recipient diagnosed with any congenital abnormalities?  
 1  yes  
 2  no

Specify test(s) performed to identify disease:

9. 1  yes 2  no Abnormal facies (snub nose, thick upper lip, epicanthic folds, hypertelorism)

10. 1  yes 2  no Ear abnormalities

11. 1  yes 2  no Eye abnormalities (strabismus, cataract, microphthalmia)

12. 1  yes 2  no Other neurologic abnormalities

13. If yes, specify: \_\_\_\_\_

14. 1  yes 2  no Microcephaly

15. 1  yes 2  no Palate or jaw abnormalities (cleft palate and/or lip, Pierre Robin syndrome, small jaw or mouth)

16. 1  yes 2  no Abnormal neck (short or webbed neck)

17. 1  yes 2  no Cardiac abnormalities

18. 1  yes 2  no Exocrine pancreatic deficiency

19. 1  yes 2  no Gastrointestinal abnormalities

20. 1  yes 2  no Genital abnormalities (cryptorchism, hypoplasia)

21. 1  yes 2  no Kidney or urinary tract abnormalities

22. 1  yes 2  no Thumb abnormalities

23. 1  yes 2  no Radius abnormalities

**Mail this form to your designated campus (Milwaukee or Minneapolis). Retain the original at the transplant center.**

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24. 1  yes 2  no Other skeletal abnormalities (syndactyly, clinodactyly, abnormal ribs, metaphyseal dyschondroplasia)

25. 1  yes 2  no Cafe au lait spots

26. 1  yes 2  no Other skin abnormalities →  27. If yes, specify: \_\_\_\_\_

28. 1  yes 2  no Other congenital abnormalities →  29. If yes, specify: \_\_\_\_\_

30. Specify the date that abnormal blood results were first observed:        
 Month Day Year

31. Specify the presenting hematologic disorder:  
 1  acute leukemia → **Complete a corresponding Leukemia insert**

2  cytopenia → Specify cytopenia:  
 32. 1  yes 2  no Anemia (Hb < 10 g/dL)  
 33. 1  yes 2  no Thrombocytopenia (platelets < 100,000/mm<sup>3</sup>)  
 34. 1  yes 2  no neutropenia (ANC < 1,000/mm<sup>3</sup>)

3  myelodysplasia → **Complete a Form 2014 – MDS insert**

4  other disorder → 35. Specify other hematologic disorder: \_\_\_\_\_

36. Was the bone marrow examined at diagnosis?  
 1  yes →  
 2  no  
 3  unknown

37. Specify date of bone marrow examination:        
 Month Day Year

38. Specify cellularity:  
 1  decreased  
 2  normal  
 3  increased  
 4  unknown

39. Were dysplastic features present at diagnosis?  
 1  yes  
 2  no  
 3  unknown

40. Blasts in marrow:    %

### Cytogenetic Studies / Sensitivity to DNA Cross-Linking Agents

This section requests information about bone marrow karyotype and examination of bone marrow and blood cells for non-specific chromatid abnormalities. If more than one investigation was carried out, please copy this section and report each event separately.

41. Was bone marrow karyotyping performed at diagnosis?  
 1  yes →  
 2  yes, but no evaluable metaphases  
 3  no  
 4  unknown

42. Was karyotype normal?  
 1  yes  
 2  no → Specify abnormalities identified:

43. 1  yes 2  no +4  
 44. 1  yes 2  no +5  
 45. 1  yes 2  no -7  
 46. 1  yes 2  no +8  
 47. 1  yes 2  no +11



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79. Was complementation group testing performed at any time prior to the preparative regimen?

- 1  yes  
2  no

Specify groups identified:

80. 1  yes 2  no FANCA  
81. 1  yes 2  no FANCB  
82. 1  yes 2  no FANCC  
83. 1  yes 2  no FANCD2  
84. 1  yes 2  no FANCE  
85. 1  yes 2  no FANCF  
86. 1  yes 2  no FANCG  
87. 1  yes 2  no Other group → 88. If yes, specify:

89. Is a copy of the complementation group report attached?  
1  yes  
2  no

90. Were any genetic mutations identified?

- 1  yes  
2  no

Specify mutation origin:

91. 1  yes 2  no maternal → **Copy questions 93–104 and complete for mother**  
92. 1  yes 2  no paternal → **Copy questions 93–104 and complete for father**

93. Was a mutation analysis of cloned Fanconi Anemia genes performed at any time prior to the preparative regimen?

- 1  yes  
2  no  
3  unknown

94. Specify the date the analysis was performed:     
Month Day Year

Specify mutation(s): Specify value:

95. 1  yes 2  no Exon → 96. Specify:

97. 1  yes 2  no Intron → 98. Specify:

99. 1  yes 2  no Nucleotide change (e.g., 732G>C) → 100. Specify:

101. 1  yes 2  no Amino acid changes (e.g., L244F) → 102. Specify:

103. Specify the mutation type:  
1  substitution  
2  deletion  
3  insertion

104. Is a copy of the mutation analysis report attached?  
1  yes  
2  no

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105. Were the recipient's bone marrow cells or peripheral blood mononuclear cells tested for sensitivity to cross-linking agents?

- 1  yes
- 2  no
- 3  unknown

106. Specify the date the testing was performed:     
Month Day Year

107. Specify the type of cross-linking agent used:  
1  diepoxybutane (DEB)  
2  mitomycin C  
3  other agent ➤ 108. Specify other agent:

109. Were chromatid aberrations present on an *unstressed* preparation?  
1  yes  
2  no  
3  not evaluable

110. Total number of cells studied:   
111. Number of aberrations per cell:   
112. Number of cells with no aberrations:

113. Were chromatid aberrations present on a *stressed* preparation?  
1  yes  
2  no  
3  not evaluable

114. Total number of cells studied:   
115. Number of aberrations per cell:   
116. Number of cells with no aberrations:

117. Is a copy of the report attached?  
1  yes  
2  no

### Familial History of Disease

118. Were any other genetically related family members affected?

- 1  yes
- 2  no
- 3  unknown

Specify family member(s):

119. 1  yes 2  no Sibling  
120. 1  yes 2  no Cousin  
121. 1  yes 2  no Parent  
122. 1  yes 2  no Aunt / uncle  
123. 1  yes 2  no Other relative ➤ 124. Specify relationship:

125. Is the recipient genetically related to his / her parents?

- 1  yes, both mother and father
- 2  yes, mother only
- 3  yes, father only
- 4  no, not genetically related to mother or father
- 5  unknown

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126. (Related donors only) Were the donor's blood or bone marrow cells tested for sensitivity to cross-linking agents?

- 1  yes
- 2  no
- 3  unknown

127. Specify the date testing was performed:     
Month Day Year

128. Specify the type of cross-linking agent used:  
1  diepoxybutane (DEB)  
2  mitomycin C  
3  other agent ➤ 129. Specify other agent:

130. Were chromatid aberrations present on an *unstressed* preparation?  
1  yes  
2  no  
3  not evaluable

131. Total number of cells studied:   
132. Number of aberrations per cell:   
133. Number of cells with no aberrations:

134. Were chromatid aberrations present on a stressed preparation?  
1  yes  
2  no  
3  not evaluable

135. Total number of cells studied:   
136. Number of aberrations per cell:   
137. Number of cells with no aberrations:

138. Is a copy of the report attached?  
1  yes  
2  no

139. Was the recipient treated with androgens prior to the HSCT?

- 1  yes
- 2  no
- 3  unknown

140. Was the recipient treated with corticosteroids prior to the HSCT?

- 1  yes
- 2  no
- 3  unknown

141. Did the recipient receive growth factors prior to the HSCT?

- 1  yes
- 2  no
- 3  unknown

If yes, specify cytokine(s) given:

142. 1  yes 2  no Erythropoietin (includes all forms of erythropoietin / darbepoetin)

143. 1  yes 2  no G-CSF

144. 1  yes 2  no GM-CSF

145. 1  yes 2  no IL3

146. 1  yes 2  no Neulasta

147. 1  yes 2  no Stem cell factor

148. 1  yes 2  no Other growth factor ➤ 149. Specify other agent:

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150. Did the recipient receive red blood cell transfusions between diagnosis and the start of the preparative regimen?

- 1  yes  
2  no

151. Specify the total number of donor exposures (best estimate):

- 1  1-5  
2  6-10  
3  11-20  
4  21-30  
5  31-40  
6  41-50  
7  ≥ 51  
8  unknown

152. Did the recipient receive platelet transfusions in the four weeks prior to the preparative regimen?

- 1  yes  
2  no

### Clinical Features Just Prior to the Preparative Regimen

153. What was the recipient's disease status immediately prior to the preparative regimen?

- 1  stable cytopenia, no cytogenetic abnormalities (no MDS)  
2  stable cytopenia with cytogenetic abnormalities (no MDS)  
3  progressive cytopenia  
4  myelodysplasia → **Complete a Form 2014 – MDS insert**  
5  leukemia, untreated → **Complete a corresponding Leukemia insert**  
6  leukemia, treated → **Complete a corresponding Leukemia insert**

### Hematologic Parameters Immediately Prior to the Preparative Regimen

154. Was the recipient's bone marrow examined at any time between diagnosis and the preparative regimen?

- 1  yes  
2  yes, but no  
    evaluatable  
    metaphases  
3  no  
4  unknown

155. Were any karyotype abnormalities identified?

- 1  yes  
2  no

Specify abnormalities identified:

- |      |                                |                               |                     |
|------|--------------------------------|-------------------------------|---------------------|
| 156. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | +4                  |
| 157. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | +5                  |
| 158. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | -7                  |
| 159. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | +8                  |
| 160. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | +11                 |
| 161. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | +13                 |
| 162. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | +14                 |
| 163. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | -17                 |
| 164. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | -18                 |
| 165. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | -20                 |
| 166. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | +21                 |
| 167. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | +22                 |
| 168. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | -X                  |
| 169. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | -Y                  |
| 170. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | del(5q)             |
| 171. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | del(7q)             |
| 172. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | del(9q)             |
| 173. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | del(11q)            |
| 174. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | del(20q)            |
| 175. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | inv(3) or t(3;3)    |
| 176. | 1 <input type="checkbox"/> yes | 2 <input type="checkbox"/> no | inv(16) or t(16;16) |

